Office of Genomics and Disease Prevention

The Office of Genomics and Disease Prevention (OGDP) was established in 1997 and has since provided national leadership while building partnerships with other federal agencies, public health organizations, professional groups, and the private sector. The mission of OGDP is to integrate genomics into public health research, policy, and programs and to improve population health and prevent disease through the application of genomic information

Top priorities

- Integrating genomics into public health research.
- Assessing the value of family history and genomic tests for population health.
- Incorporating genomics into public health practice.

Major Accomplishments

Integrating Genomics into Public Health Research

Acute Public Health Investigations
OGDP formed a multidisciplinary Acute Public Health Investigation (APHI) Working Group in collaboration with the Council of State and Territorial Epidemiologists (CSTE) to develop a plan and tools for incorporating genomics into APHIs. A workshop convened on May 12-13, 2004 addressed "*The Role of Human Genomics in Acute Public Health Investigations: Current Practice and Future Strategies."* Workshop participants reviewed the rationale, state of the science, challenges and opportunities for incorporating human genomics into acute public health investigations. They outlined next steps for prioritizing investigations and developing the necessary capacity at CDC to incorporate human genomics. Input from the workshop will inform a new initiative to develop criteria for incorporating human genomics into APHIs and develop standard tools and protocols for field and laboratory workers.

NHANES Working Group
In collaboration with the National Institutes of Health (NIH), a CDC-wide team is measuring population variation in selected genes using stored DNA samples collected during the third National Health and Nutrition Examination Survey (NHANES) III. The goal of this collaboration is to develop genotype prevalence estimates based on a nationally representative sample of the U.S. population. These data will add another dimension to the analysis of clinical, physical, and lifestyle information collected by NHANES, creating a resource for analysis of genotype-phenotype correlations and gene-environment interactions.

HuGENet

OGDP established the Human Genome Epidemiology Network (HuGeNet™, http://www.cdc.gov/genomics/hugenet) as a global collaboration of individuals and organizations committed to assessing the role of human genome variation in population health and the potential of genomics for improving health and preventing disease. HuGENet promotes publication of systematic reviews of population-based data on genotype prevalence, gene-disease associations, and gene-environment interactions. Accomplishments in 2004 include publication of a book, *Human Genome Epidemiology*, and co-sponsorship of a workshop on meta-analysis by the Public Health Genetics Unit in Cambridge, UK (http://www.cgkp.org.uk/work/activities.html#syst_rev).

Assessing the value of family history and genomic tests for population health

Family History Public Health Initiative

The *Family History Public Health Initiative* (http://www.cdc.gov/genomics/info/factshts/famhist.htm), launched in 2002, has produced a new family history tool, Family Healthware™, which is a web-based and self-administered. It collects information on family health history and personal risk factors. It calculates familial risk using algorithms built into the software and generates a report that includes a family health pedigree and tailored prevention messages and screening recommendations. A resource manual being developed for healthcare providers will include an explanation of familial risk levels and possible genetic conditions underlying high risk, recommendations for further risk assessment, and suggested interventions and resources. In 2005, the clinical utility of Family Healthware™ will be evaluated in a randomized controlled trial conducted by three academic medical centers.

On November 8th, 2004 the Surgeon General held a national press conference to encourage Americans to make Thanksgiving Day the first annual National Family History Day and to direct them to the Surgeon General's web site, which includes *My Family Health Portrait,* an abbreviated version of the CDC family history tool. OGDP has also launched a new family history website for the general public that includes fact sheets, case studies, news articles, and presentations.

Evaluation of Genetic Testing

In fall 2004, OGDP launched *Evaluation of Genomic Applications in Practice and Prevention* (*EGAPP),*a new project whose goal is to develop and evaluate a coordinated process for systematic assessment of genetic tests in transition from research to clinical and public health practice in the U.S.. A key element for achieving success in this project will be the development and maintenance of partnerships and collaborations with stakeholders (e.g., US Preventive Services Task Force, CDC Community Guide), other HHS agencies, the international health technology assessment community, and other relevant projects and advisory groups.

On September 27-28th, 2004, OGDP held a short course, *Public Health Assessment of Genetic Tests for Screening and Prevention,* on systematic approaches to evidence-based assessment of genetic tests. Participants included public health and health care professionals, policymakers and other who are involved in the development, evaluation, utilization and reimbursement of genetic tests for screening and prevention.

Incorporating genomics into public health practice

Training Public Health Professionals in Genomics

The Michigan Center for Genomics and Public Health, in collaboration with Genomics Centers at the University of North Carolina and University of Washington, OGDP, and the Chronic Disease Directors, completed a web-based training program for public health professionals called *Six Weeks to Genomic Awareness*. The program is available as a series of six presentations designed to help public health professionals understand how genomic advances are relevant to public health. The series introduces the user to genomic concepts such as the human genome and heredity, genetic variation in populations, genetic testing, and ethical, legal, and social issues from a public health perspective. It is capped off by an overview of state and national resources for public health professionals.

OGDP and the three Centers for Genomics and Public Health also created *Genomics for Public Health Practitioners,* a 45-minute introductory presentation for public health practitioners. It describes the application of genomics to public health, dispels some myths and identifies challenges.

Future Directions

1. Develop a CDC-wide plan to enhance capacity in public health genomics research, evidence-based assessment and integration of genomics into practice.

2. Develop a public-private initiative to use NHANES DNA bank for intramural and extramural public health genomics research.

3. Launch the Evaluation of Genomic Applications in Practice and Prevention (EGAPP) initiative.

4. Develop and implement guidance for the systematic evaluation of gene-disease associations.

5. Develop guidance for the systematic design and reporting of results from genomic cohort ("biobank") studies.

6. Conduct a national workshop on methods for applying family history tools in preventive medicine and public health.

Publications

Khoury MJ, Millikan R, Little J, Gwinn M. The emergence of epidemiology in the genomics age. Int J Epidemiol 2004;33:936-44.

McCusker ME, Yoon PW, Gwinn M, Malarcher AM, Neff L, Khoury MJ. Family history of heart disease and cardiovascular disease risk-reducing behaviors. Genet Med 2004;6:153-8.

Khoury MJ, Yang Q, Gwinn M, Little J, Dana Flanders W. An epidemiologic assessment of genomic profiling for measuring susceptibility to common diseases and targeting interventions. Genet Med 2004;6:38-47.

Scheuner MT, Yoon PW, Khoury MJ. Contribution of Mendelian Disorders to Common Chronic Disease: Opportunities for Recognition, Intervention, and Prevention. *Am J Med Genet* 2004;125C:50-65.

Lindegren ML, et al. Applying public health strategies to primary immunodeficiency disorders: a model approach to genetic disorders. MMWR 2004 (RR01);53:1-29.

Khoury MJ. The case for a global human genome epidemiology initiative. Nat Genet 2004;36:1027-1028.

Yoon, PW, Scheuner MT, Gwinn M, Khoury MJ, Jorgensen C, Hariri S, Lyn S. Awareness of family health history as a risk factor for disease, United States, 2004. MMWR 2004; 53(44):1044-1047

Myers, M, Jorgensen CJ, et al. Genetic Testing for Breast and Ovarian Cancer Susceptibility: Evaluating Direct-to-Consumer Marketing—Atlanta, Denver, Raleigh-Durham, and Seattle, 2003. MMWR July;53(27):603-606.

Human Epidemiology: A Scientific Foundation for Using Genetic Information to Improve Health and Prevent Disease was published by Oxford University Press in 2004. It provides a scientific foundation to help researchers, policy makers, and practitioners integrate genomics into medical and public health practice.

Publication of the first annual OGDP report titled, "*Genomics and Population Health: United States 2003*." The report includes a timely and practical collection of vignettes of the status of genomics and population health in the United States, and is intended for public health professionals who are interested in integrating genomics into health promotion, disease prevention and healthcare.

Publications from the ACCE Project included *Clinical sensitivity of prenatal screening for cystic fibrosis via CFTR carrier testing in a United States panethnic population* (Palomaki et al., Genetics in Medicine 6:405-414), and *Prenatal screening for cystic fibrosis: An early report card (editorial)* (Palomaki, Genetics in Medicine 6:115). The American College of Medical Genetics utilized data from the ACCE review on cystic fibrosis carrier testing to revise recommendations for the mutation panel (Watson et al, *Cystic fibrosis population carrier screening: 2004 revision of American College of Medical Genetics mutation panel.* Genetics in Medicine 6:387-91). Papers in press at the end of 2004 were *An evaluation of BRCA1 and BRCA2 founder mutations penetrance estimates for breast cancer among Ashkenazi Jewish women* and *Adjusting the estimated proportion of breast cancer cases associated with BRCA1 and BRCA2 mutations: Public health implications* (McClain et al., Genetics in Medicine).